SERPINE1 gene

serpin family E member 1

Normal Function

The *SERPINE1* gene provides instructions for making a protein called plasminogen activator inhibitor 1 (PAI-1). PAI-1 is involved in normal blood clotting (hemostasis). After an injury, clots protect the body by sealing off damaged blood vessels and preventing further blood loss.

The PAI-1 protein blocks (inhibits) the action of other proteins called plasminogen activators. These proteins, including urokinase plasminogen activator (u-PA) and tissue type plasminogen activator (t-PA), convert an inactive enzyme called plasminogen to its active form, plasmin. Plasmin is involved in fibrinolysis, which is the process of dissolving blood clots. By inhibiting the conversion of plasminogen to plasmin, and thereby preventing fibrinolysis, the PAI-1 protein helps ensure that clots are only dissolved when they are no longer needed to stop bleeding.

In addition to its role in hemostasis, PAI-1 is also thought to be involved in cell movement (migration) and the breakdown and replacement (remodeling) of body tissues.

Health Conditions Related to Genetic Changes

Complete plasminogen activator inhibitor 1 deficiency

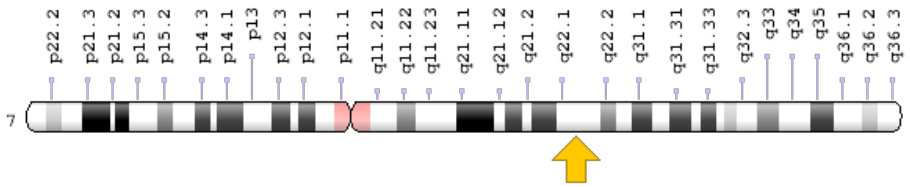
At least three *SERPINE1* gene mutations have been identified in people with complete plasminogen activator inhibitor 1 (PAI-1) deficiency, a disorder that causes abnormal bleeding. In people with this disorder, bleeding after an injury can be excessive and last longer than usual. Affected females may have excessive bleeding associated with menstruation (menorrhagia), and abnormal bleeding in pregnancy and childbirth.

The *SERPINE1* gene mutations that cause complete PAI-1 deficiency result in impaired production of the PAI-1 protein, or lead to production of PAI-1 protein that is nonfunctional or that is unstable and quickly broken down. Absence of functional PAI-1 protein allows plasminogen activators to dissolve blood clots prematurely, resulting in the abnormal bleeding associated with this disorder.

Chromosomal Location

Cytogenetic Location: 7q22.1, which is the long (q) arm of chromosome 7 at position 22.1

Molecular Location: base pairs 101,127,104 to 101,139,247 on chromosome 7 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- endothelial plasminogen activator inhibitor
- PAI
- PAI-1
- PAI1
- PLANH1
- plasminogen activator inhibitor 1 precursor
- serine (or cysteine) proteinase inhibitor, clade E (nexin, plasminogen activator inhibitor type 1), member 1
- serpin E1
- serpin peptidase inhibitor, clade E (nexin, plasminogen activator inhibitor type 1), member 1

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): The Clotting Process Must Be Precisely Regulated
  https://www.ncbi.nlm.nih.gov/books/NBK22589/#A1412

Clinical Information from GeneReviews

- Complete Plasminogen Activator Inhibitor 1 Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK447152

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SERPINE1%5BTIAB%5D%29+OR+%28serpin+family+E+member+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SERPIN PEPTIDASE INHIBITOR, CLADE E (NEXIN, PLASMINOGEN ACTIVATOR INHIBITOR TYPE 1), MEMBER 1
  http://omim.org/entry/173360

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SERPINE1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SERPINE1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:8583
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5054
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5054
- UniProt
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